

POLYCYSTIC KIDNEY DISEASE PKD2 GENE AND USES THEREOF

Abstract of the Disclosure

The present invention provides a purified and isolated wild type *PKD2* gene, as well as mutated forms of this gene. The present invention also provides one or more single-stranded nucleic acid probes which specifically hybridize to the wild type *PKD2* gene or the mutated *PKD2* gene, and mixtures thereof, which may be formulated in kits, and used in the diagnosis of ADPKD associated with the mutated *PKD2* gene. The present invention also provides a method for diagnosing ADPKD caused by a mutated *PKD2* gene, as well as a method for treating autosomal dominant polycystic kidney disease caused by a mutated *PKD2* gene.

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